Letter to the Editor

Regarding Trisomy 2p Syndrome

To the Editor:

We read with interest the publication by Hahm et al. [1999], "Trisomy 2p syndrome: A fetus with anencephaly and postaxial polydactyly." We would like to report on another case of anencephaly in a fetus with triplication of 2p23.1→2pter resulting from a paternally derived translocation. The karyotype in this case was 46,XX,der(3)t(2;3)(p23.1;q29)pat.

The fetus was diagnosed at 15 weeks with anencephaly when the mother presented for amniocentesis on account of the known paternal translocation. Autopsy confirmed anencephaly and demonstrated, in addition, a sacral meningomyelocoele; no other abnormalities were detected.

A paternal sister with the same unbalanced translocation is now 25 years old. She was not available for examination but is said to be severely retarded with absent tear ducts and a scoliosis. She is able to walk.

Neural tube defects have now been reported in several cases with trisomy 2p [Fineman et al., 1983; Sarda et al., 1992; Singer and Gerson, 1987; Thirkelsen et al., 1973; Winsor et al., 1997]. The triplicated segments consisted of 2p23→2pter, 2p13→2pter, 2p24→2pter, 2p13→2pter, and 2p13→2p24, respectively. As in case and that of Hahm et al., all included 2p24.

The case presented here adds further support to the suggestion by Lurie et al. [1995] and Winsor et al. [1997] that 2p24 is a candidate region for genes responsible for neural tube development.

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Diana Wellesley*
Wessex Clinical Genetics Service
Princess Anne Hospital
Southampton, United Kingdom

Tracy Boyle
Wessex Regional Genetics Laboratory
Salisbury District Hospital
Salisbury, United Kingdom

*Correspondence to: Dr. Diana Wellesley, Wessex Clinical Genetics Service, Princess Anne Hospital, Coxford Road, Southampton S016 5YA, U.K.

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